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Revolutionizing cfDNA testing with self-collected capillary blood samples

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Objective

Non-invasive prenatal screening (NIPS) has transformed prenatal care, but phlebotomy requirements and high test costs have hindered universal adoption, leading to inequitable access to cell-free DNA screening (cfDNA). This study evaluated a novel NIPS method using self-collected capillary blood samples to address these challenges.

Methods

We conducted an IRB-approved comparative study inclusive of venous and capillary blood samples from over 200 pregnant individuals to establish NIPS assay performance. Additionally, we analyzed the first 2000 self-collected capillary blood samples from clinical practice at our commercial NIPS laboratory. Screening performance was assessed by comparing capillary and venous samples' sensitivity and specificity for common aneuploidy detection and accuracy in fetal sex distinction.

Results

Our comparative study showed >99% sensitivity and >99.9% specificity for autosomal trisomy detection, and >97% sensitivity and >99.9% specificity for sex chromosome aneuploidies. In the clinical evaluation of the first 2000 capillary blood samples, the novel method demonstrated >99% accuracy for fetal sex distinction.

Conclusion

Our novel NIPS method utilizing self-collected capillary blood shows high sensitivity and specificity in aneuploidy screening and high accuracy in fetal sex distinction; therefore, our method represents a potential paradigm shift in the accessibility of prenatal screening. To date, NIPS has required phlebotomy, without a self-collection option through saliva or buccal samples such as may be used with other molecular genetic tests. Self-collection of capillary blood is convenient, cost-efficient, and offers a viable approach for NIPS, ultimately promoting equitable access to cell-free DNA screening. The widespread adoption and integration of this novel method into clinical practice can enhance NIPS accessibility and associated outcomes for expectant parents, particularly those in underserved populations. Our findings demonstrate the potential for self-collected capillary blood to improve NIPS accessibility and promote equitable access to this important component of prenatal care.